National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions

September 2014

GenTAC Clinical Center Highlight: Oregon Health & Science University

In this issue we highlight Oregon Health & Science University, a nationally prominent research university and Oregon's only public academic health center. Its hospitals and clinics serve more than a quarter of a million patients every year with innovative care and treatment models based on the latest knowledge available. Cheryl Maslen, PhD is the GenTAC Principal Investigator at OHSU. Dr. Maslen is a professor of cardiovascular medicine and molecular and medical genetics and the associate director of the OHSU Heart Research Center. Her co-Investigators for GenTAC are Dr. Michael Silberbach, MD, professor of pediatric cardiology and pediatric cardiologist with a special interest in problems of the aorta in Turner syndrome and Dr. Howard Song, MD, PhD associate professor on the faculty of the Division of Cardiothoracic Surgery at OHSU. The GenTAC study coordinator is Alisha Berry who has worked in research at OHSU for two years. She is in the process of applying to medical schools and is currently hoping to pursue a specialty in pediatric endocrinology.

Turner Syndrome Society of the United States (TSSUS) Annual Conference

- In July, GenTAC and Oregon Health & Science University attended the annual TSSUS conference. We have attended this conference since 2008 to enroll and follow patients in the GenTAC registry. OHSU met with almost 70 subjects to collect updated information and perform follow-ups. This conference is always such a great opportunity to connect with the subjects and the team at OHSU is already looking forward to next year's conference in Kansas City, MO!

3rd Thoracic Aortic Disease Summit

- GenTAC hosted the Third Thoracic Aortic Disease Summit in Baltimore, MD on July 9-10, 2014. Over 130 experts in the fields of cardiovascular care, surgery and research, from the US and abroad, attended this meeting to share their current research, exchange ideas about diagnosis, treatment and management. Catherine Boileau, PhD and Guillaume Jondeau, MD were presented with the Aortic Disease Summit Award for recognition of recent outstanding contributions elucidating genetic aortic diseases. Plans are underway for the 4th Summit in 2016. Please visit **https://gentac.nhlbi.nih.gov/AorticSummit.aspx** for updates!

Thoracic Aortic Disease Summit

Left to right: Dianna M. Milewicz, Catherine Boileau, Guillaume Jondeau and Kim Eagle

Steering Committee Members

Principal Investigators	
Scott A. LeMaire, MD	Baylor College of Medicine
William Ravekes, MD	Johns Hopkins University School of Medicine
Cheryl L. Maslen, PhD	Oregon Health & Science University
Ralph V. Shohet, MD	Queen's Medical Center
Reed E. Pyeritz, MD, PhD	University of Pennsylvania School of Medicine
Dianna M. Milewicz, MD, PhD	University of Texas Medical School at Houston
Richard B. Devereux, MD	Weill Cornell Medical College of Cornell University
Core Labs	
Jennifer P. Habashi, MD	Johns Hopkins University Hospital
Federico M. Asch, MD	MedStar Research Institute
Data Coordinating Center	
Barbara L. Kroner, PhD	RTI International
NHLBI	
H. Eser Tolunay, PhD	National Heart, Lung, and Blood Institute
SC Chair	
Kim A. Eagle, MD	University of Michigan

Other Recent Updates

- 8 **videocasts** of the GenTAC investigators discussing the research they have done using the GenTAC Registry are available on the GenTAC website. View Scott LeMaire, M.D., Professor of Surgery and of Molecular Physiology and Biophysics at Baylor College of Medicine highlights the controversy about treating aortic valve regurgitation in MFS patients undergoing ascending aortic repair. New videocasts will be released later this year!
- 4 GenTAC abstracts have been accepted at upcoming meetings:
  - » Parkash, Siddharth. Common autosomal variants are associated with bicuspid aortic valve in Turner Syndrome. American Heart Association, November 15–19, 2014
  - » Doyle, Jeff. Calcium Channel Blockers Accelerate Aortic Aneurysm and Cause Premature Lethality in Marfan Syndrome and Related Conditions. American Heart Association, November 15–19, 2014
  - » Guo, Dongchuan. MAT2A Mutations Cause Familial Thoracic Aortic Aneurysms and Aortic Dissections. American Society of Human Genetics, October 18–22, 2014
  - » Oswald, Gretchen, Updated cardiac description in Loeys Dietz syndrome, at ASHG in October

Do you have a research interest in genetically triggered thoracic aortic conditions?

GenTAC makes its collection of medical data and biologic samples available at no cost to qualified investigators. Your work can help determine best practices that advance the clinical management of genetic aortic aneurysms and other cardiovascular conditions.

To submit a proposal to use GenTAC data or for more information, visit our **website: https://gentac.nhlbi.nih.gov/**.

A Snapshot of Who is Enrolled in GenTAC

Number of people enrolled: 3763

Biospecimens

Blood:	2114
Saliva:	1361
Both blood and saliva	93
Tissue:	151

Gender

Male:	2221
Female:	1527

» To submit a proposal to use GenTAC data or for more information, visit our website: **https://gentac.nhlbi.nih.gov/.**

Eligible Diagnosis

Marfan:	897
Turner:	314
Ehlers-Danlos (vascular): other):	164
Ehlers-Danlos (other):	27
Loeys-Dietz:	104
FBN1, TGFBR mutation:	44
BAV with aortic enlargement:	930
BAV with family history:	23
BAV with coarctation:	83
Shprintzen-Goldberg:	5
Familial TAA:	276
Other aneurysm, dissections:	744
Other congenital heart disease:	113

Age

<5	128
5 - 17	653
18-39	1142
40-69	1712
>69	112

Race

White, non-Hispanic	3035
Black, non-Hispanic	190
Hispanic	289
Asian	148
American Indian/Alaskan native	19
Native Hawaiian/Pac. Islander	69